



Introducing Rare Diseases

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Introducing *Rare Diseases*

Patricia Szajner¹ and Timur Yusufzai²

¹Landes Bioscience; Boston, MA USA; ²Harvard Medical School; Boston, MA USA

We are pleased to introduce *Rare Diseases*, an open access journal dedicated to publishing high-quality research that addresses the many aspects related to rare diseases. *Rare Diseases* will cover a range of topics, including the studies of disease-related proteins, the analyses of rare disease mutations, gene expression studies, genotype-phenotype correlations, studies using animal models, novel clinical findings and advances in rare disease therapeutics. To achieve this mission, *Rare Diseases* relies on an exceptional Editorial Board comprised of internationally recognized leaders in their fields. The diverse background of the Editorial Board mirrors the diversity of topics that will be covered by *Rare Diseases*.

The launching of *Rare Diseases* comes as research into the genetics and therapeutics of rare diseases intensifies. There are approximately 7,000 rare diseases and it is estimated that they affect almost 10% of the population in the United States (US).^{1,2} In the US, a disease or disorder is typically defined as rare when it affects less than 200,000 people at any given time. In Europe, a disease is labeled rare when it affects less than one in 2,000 people. The advent of genome-wide sequencing studies have accelerated the discovery of disease-causing mutations and facilitated research into the underlying mechanisms of different diseases. Research on rare diseases not only provides essential insight into human diseases, but also provides invaluable understanding of normal cellular processes.

In the past 30 years, there have been many efforts to increase research and awareness on rare diseases.² The National Organization for Rare Disorders (NORD) had a strong influence on the passage of the Orphan Drug Act of 1983, which has helped spur the development of more than 400 therapeutics for rare diseases. The National Institutes of Health created the Office of Rare Disease Research (ORDR), which was established in the Rare Disease Act of 2002. The ORDR is tasked with supporting rare disease research and providing information on rare diseases. With the increase in research on rare diseases and the advances in orphan drug development, we feel it is important to create a centralized journal on rare diseases. We believe *Rare Diseases* will fulfill that need and help facilitate continued research on rare diseases.

We chose to publish *Rare Diseases* as an open access journal. We believe that it is important to make our reports freely available not only to the scientific community, but also to patients, families, foundations, advocacy groups and anyone interested in learning about rare diseases. *Rare Diseases* will publish a variety of articles including original research manuscripts, reviews, addenda and discussions about rare disease diagnoses. From time to time, *Rare Diseases* will also include highlights from foundations and patient organizations dedicated to *Rare Diseases*. We hope this will raise awareness of various rare diseases as well as facilitate the communication of available resources such as reagents, cell lines, sequencing data or funding opportunities.

We hope that with the support of those involved in rare disease research we will be successful in providing a resource for the community.

References

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Correspondence to: Patricia Szajner and Timur Yusufzai; Email: patricia@landesbioscience.com and tyusufzai@lroc.harvard.edu

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